



COL3A1 gene

collagen type III alpha 1 chain

Normal Function

The *COL3A1* gene provides instructions for making type III collagen. Collagens are a family of proteins that strengthen and support many tissues in the body. Type III collagen is found in the skin, lungs, intestinal walls, and the walls of blood vessels.

The components of type III collagen, called pro- α 1(III) chains, are produced from the *COL3A1* gene. Each molecule of type III procollagen is made up of three copies of this chain.

The triple-stranded, rope-like procollagen molecules are processed by enzymes outside the cell to create mature type III collagen. The collagen molecules then arrange themselves into long, thin fibrils that form stable interactions (cross-links) with one another and with other types of collagen in the spaces between cells. The cross-links result in the formation of very strong collagen fibers.

Health Conditions Related to Genetic Changes

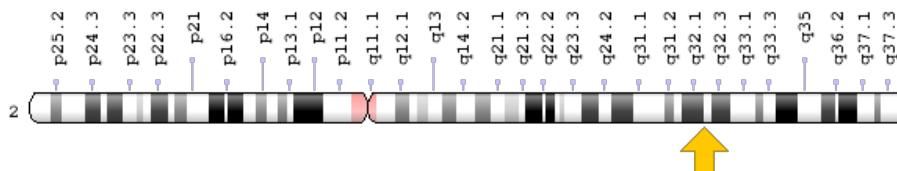
Ehlers-Danlos syndrome

More than 500 mutations in the *COL3A1* gene have been found to cause a form of Ehlers-Danlos syndrome called the vascular type. Ehlers-Danlos syndrome is a group of disorders that affect the connective tissues that support the skin, bones, blood vessels, and many other organs and tissues. The vascular type can cause potentially life-threatening complications, including tearing (rupture) of blood vessels, intestines, and other organs. The mutations that cause this form of the disorder alter the structure and production of type III procollagen molecules. As a result, a large percentage of type III collagen molecules are assembled incorrectly, or the amount of type III collagen is greatly reduced. Researchers believe that these changes affect tissues that are normally rich in this type of collagen, such as the skin, blood vessel walls, and internal organs. An insufficient amount of type III collagen weakens connective tissues in these parts of the body, causing the signs and symptoms of the vascular type of Ehlers-Danlos syndrome.

Chromosomal Location

Cytogenetic Location: 2q32.2, which is the long (q) arm of chromosome 2 at position 32.2

Molecular Location: base pairs 188,974,373 to 189,012,746 on chromosome 2 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- alpha 1 type III collagen
- CO3A1_HUMAN
- collagen III, alpha-1 polypeptide
- collagen type III alpha 1
- collagen, fetal
- collagen, type III, alpha 1
- collagen, type III, alpha 1 (Ehlers-Danlos syndrome type IV, autosomal dominant)
- EDS4A

Additional Information & Resources

Educational Resources

- Molecular Cell Biology (fourth edition, 2000): Collagen: The Fibrous Proteins of the Matrix
<https://www.ncbi.nlm.nih.gov/books/NBK21582/>

GeneReviews

- Vascular Ehlers-Danlos Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1494>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28COL3A1%5BTIAB%5D%29+OR+%28collagen%5BTI%5D%29+AND+%28type+III%5BTI%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

OMIM

- COLLAGEN, TYPE III, ALPHA-1
<http://omim.org/entry/120180>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_COL3A1.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=COL3A1%5Bgene%5D>
- Ehlers-Danlos Syndrome Variant Database
https://eds.gene.le.ac.uk/home.php?select_db=COL3A1
- HGNC Gene Family: Collagens
<http://www.genenames.org/cgi-bin/genefamilies/set/490>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2201
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/1281>
- UniProt
<http://www.uniprot.org/uniprot/P02461>

Sources for This Summary

- Beridze N, Frishman WH. Vascular Ehlers-Danlos syndrome: pathophysiology, diagnosis, and prevention and treatment of its complications. *Cardiol Rev.* 2012 Jan-Feb;20(1):4-7. doi: 10.1097/CRD.0b013e3182342316. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/22143279>
- Germain DP. Ehlers-Danlos syndrome type IV. *Orphanet J Rare Dis.* 2007 Jul 19;2:32. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/17640391>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1971255/>
- Giunta C, Steinmann B. Characterization of 11 new mutations in COL3A1 of individuals with Ehlers-Danlos syndrome type IV: preliminary comparison of RNase cleavage, EMC and DHPLC assays. *Hum Mutat.* 2000 Aug;16(2):176-7.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/10923041>

- Oderich GS, Panneton JM, Bower TC, Lindor NM, Cherry KJ, Noel AA, Kalra M, Sullivan T, Gloviczki P. The spectrum, management and clinical outcome of Ehlers-Danlos syndrome type IV: a 30-year experience. *J Vasc Surg*. 2005 Jul;42(1):98-106.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/16012458>
- Schwarze U, Schievink WI, Petty E, Jaff MR, Babovic-Vuksanovic D, Cherry KJ, Pepin M, Byers PH. Haploinsufficiency for one COL3A1 allele of type III procollagen results in a phenotype similar to the vascular form of Ehlers-Danlos syndrome, Ehlers-Danlos syndrome type IV. *Am J Hum Genet*. 2001 Nov;69(5):989-1001. Epub 2001 Sep 27.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/11577371>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1274375/>

Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/gene/COL3A1>

Reviewed: November 2015

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services